Reply

Thrombosis and Risk Factors

I read the comment of Toprak et al. on my letter to the Editor appeared in a recent issue of the journal with great interest [1]. They pointed a missing point in my letter and then focused on the effect of folate metabolism on homocysteine and metihelenetetrahydrofolate gene polymorphism at 677 C to T extensively [1]

I would like to express my thanks to Toprak et al. giving a chance to explain my view on this matter once more.

As it is well known that there is a continuing debate on homocysteine metabolism, MTHFR SNP's and folate metabolism and there are several published reviews on this subject not reaching to a conclusion.

Recently we reported that MTHFR 677 T has an influence on Hcy levels in Turkish population. But also we found another possible MTHFR gene haplotype, which does not have an effect on Hcy levels [2].

Furthermore, there are also rare novel SNPs published within the MTHFR 677 region with an allele frequency of 1 in 3000-4000 sample, including MTHFR 678 C-A (Ala222Ala) in Turkish population [3-5] which may lead to erroneous technical reporting.

Since our first publication on homocysteine related gene polymorphisms in Turkish population in 1998 [6], and the others following the first paper, I reached to a conclusion that without determining homocysteine levels, analyzing MTHFR 677C-T solely at the DNA level is unnecessary, not cost effective and does not have any clinical value especially in Turkish population.

So, only homocysteine levels should be routinely analyzed and not the MTHFR 677 T SNP alone. If any cause of high homocysteine levels could not be found, then MTHFR 677 T analysis can be performed.

References

1. Akar N: Thrombosis and risk factors. Turk J Hematol 2010: 27 (4) : 318-319
2. Koç YL, Akar N: Single nucleotide polymorphisms that affect homocysteine levels in Turkish population. Clin Appl Thromb Hemost 2009; 15: 701-704
3. Lyon E: Discovering rare variants by use of melting temperature shifts seen in melting curve analysis. Clin Chem 2005; 51: 1331-1332
4. Erali M, Schmidt B, Lyon E, Wittwer C: Evaluation of electronic microarrays for genotyping factor V, factor II, and MTHFR. Clin Chem 2003; 49: 732-739
5. Eğin Y, Akar N: First observation of MTHFR 678 C-A (Ala222Ala) single nucleotide polymorphism. Turk J Hematol 2011 (DOI: 10.5152/tjh.2011.45)
6. Akar N, Akar E, Misirlioğlu M, Avcu F, Yalçin A, Cin S: Search for genetic factors favoring thrombosis in Turkish population. Thromb Res 1998; 92 (2): 79-82

Nejat Akar

TOBB Economy and Technology University Hospital, Ankara, Turkey